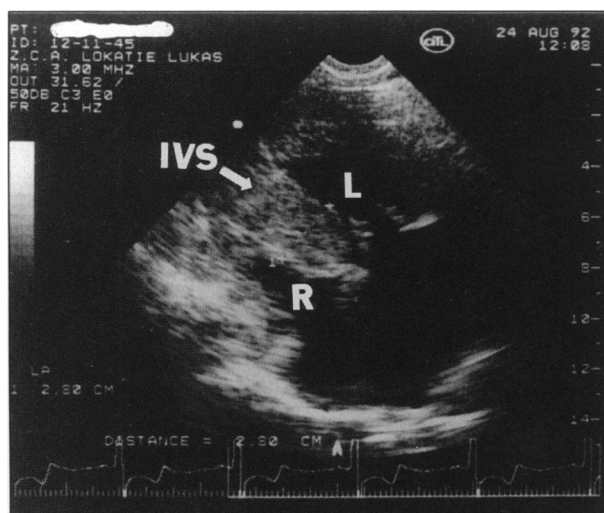
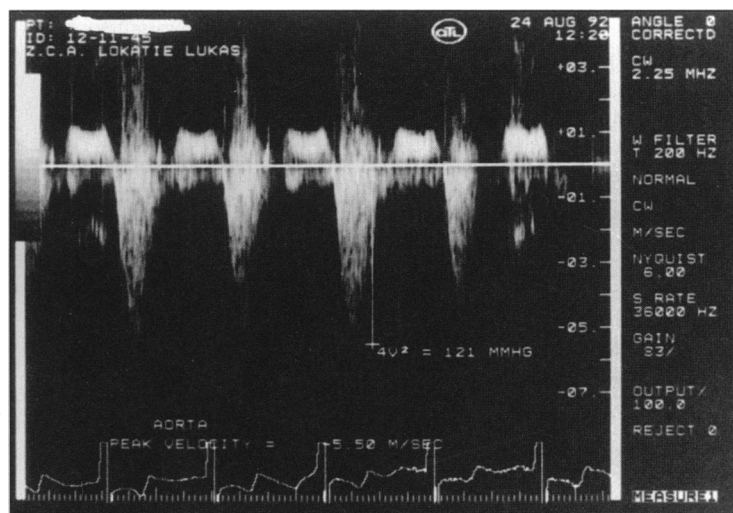


# Imaging in cardiology

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**Figure 1.** On echocardiography, the apical four-chamber view showing thickening of both the left (L) and right (R) ventricular free walls; especially the interventricular septum (IVS) is hugely thickened to 2.8 cm.



**Figure 2.** Continuous wave Doppler tracing demonstrating a peak velocity of 5.5 m/sec in the left ventricular outflow tract, which means a pressure gradient of 121 mmHg, caused by obstruction by the hugely thickened interventricular septum.

## Left ventricular outflow tract obstruction as a consequence of Sanfilippo's disease

A 46-year-old woman with Sanfilippo's disease suffering from left ventricular outflow tract obstruction

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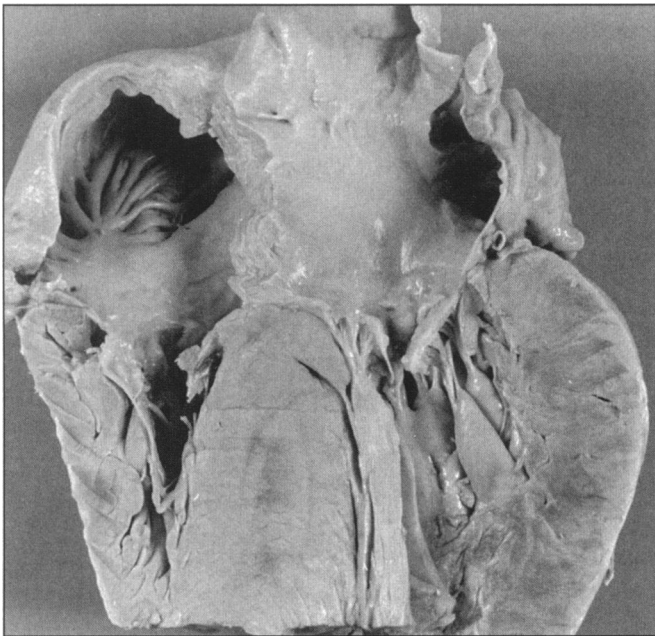
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caused by storage of heparan sulphate in the myocardium is presented here.

The patient had a history of progressive mental deterioration from the age of four years onwards, caused by Sanfilippo's disease (mucopolysaccharidosis III) type A, which is characterised by storage of heparan sulphate in the tissues. At the age of 46 years she presented with thickening of the wall of both ventricles and the interventricular septum (figure 1), causing left ventricle outflow tract obstruction (figure 2). Five years later post-mortem examination showed a thickened left ventricular wall, while septal thickening caused narrowing of the outflow tract (figure 3). Microscopic and electron-microscopic examinations (Dr Dingemans, Academic Medical Centre Amsterdam) showed storage of proteoglycans, of which heparan sulphate is one.

In the literature (1,2,3,4) we found 11 children with Sanfilippo's disease with thickening of the



**Figure 3.** Four-chamber section of the heart corresponding to the echocardiogram. Note the thickening of the free wall of the right and left ventricle. Also note the asymmetrical thickening of the ventricular septum, in this plane further accentuated by the attenuated insertion of the septal papillary muscle group.

interventricular septum. As far as we know, however, severe ventricular outflow tract obstruction due to Sanfilippo's disease has not been described in literature before. ■

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In this section a remarkable 'image' is presented and a short comment is given.

We invite you to send in images (in triplicate) with a short comment (one to two pages at the most) to Mediselect bv, Editorial Office Netherlands Heart Journal, PO Box 63, 3830 AB Leusden, the Netherlands.

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